



SATB2 gene

SATB homeobox 2

Normal Function

The *SATB2* gene provides instructions for making a protein that helps control the development of certain body systems. The SATB2 protein attaches to special regions of DNA called matrix attachment regions (MARs). These regions help determine the structure of chromatin, which is the complex of DNA and proteins that packages DNA into chromosomes. The structure of chromatin is one way that gene expression is regulated during development.

By organizing chromatin structure, the SATB2 protein coordinates the activity of multiple genes involved in the development of certain body systems. In particular, the SATB2 protein promotes the maturation of cells that build bones (osteoblasts) and directs development of structures in the head and face. The protein also plays roles in the maturation and function of different types of nerve cells (neurons) in the brain.

Health Conditions Related to Genetic Changes

SATB2-associated syndrome

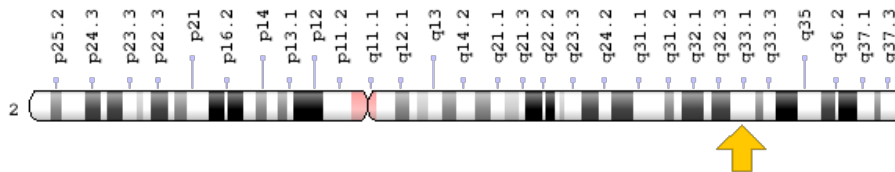
Mutations in the *SATB2* gene have been found to cause *SATB2*-associated syndrome. Individuals with this condition have intellectual disability and severe speech problems. They may also have an opening in the roof of the mouth, dental abnormalities, or other abnormalities of the head and face (craniofacial anomalies). Some of these mutations are deletions of large pieces of DNA that remove several genes, including *SATB2*. Other mutations add, remove, or rearrange smaller pieces of DNA within the *SATB2* gene. Still other mutations change single DNA building blocks (nucleotides) in the *SATB2* gene. It is likely that these genetic changes reduce the amount of functional SATB2 protein. Reduction of SATB2 function is thought to impair normal development of the brain and craniofacial structures, leading to intellectual disability, delayed speech, craniofacial anomalies, and other features of *SATB2*-associated syndrome.

The signs and symptoms of *SATB2*-associated syndrome are usually similar, regardless of the type of mutation that causes it. However, some individuals with large deletions that include additional genes have uncommon features of the condition, such as problems with the heart, genitals and urinary tract (genitourinary tract), skin, or hair. These features are thought to be related to loss of other genes near *SATB2*.

Chromosomal Location

Cytogenetic Location: 2q33.1, which is the long (q) arm of chromosome 2 at position 33.1

Molecular Location: base pairs 199,269,500 to 199,471,266 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DNA-binding protein SATB2
- FLJ21474
- GLSS
- KIAA1034
- SATB family member 2
- special AT-rich sequence-binding protein 2

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database (2000): Chromatin Mechanisms Regulating Gene Expression In Health And Disease
<https://www.ncbi.nlm.nih.gov/books/NBK45032/>
- Molecular Biology of the Cell (fourth edition, 2002): DNA Molecules Are Highly Condensed in Chromosomes
https://www.ncbi.nlm.nih.gov/books/NBK26834/#_A629_

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SATB2%5BTIAB%5D%29+OR+%28SATB+homeobox+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- SPECIAL AT-RICH SEQUENCE-BINDING PROTEIN 2
<http://omim.org/entry/608148>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SATB2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SATB2%5Bgene%5D>
- HGNC Gene Family: CUT class homeoboxes and pseudogenes
<http://www.genenames.org/cgi-bin/genefamilies/set/527>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21637
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23314>
- UniProt
<http://www.uniprot.org/uniprot/Q9UPW6>

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